SAR1B gene

secretion associated Ras related GTPase 1B

Normal Function

The *SAR1B* gene provides instructions for making a protein found in enterocytes, which are cells that line the intestine and absorb nutrients. Within enterocytes, the SAR1B protein aids in the transport of molecules called chylomicrons. As food is digested after a meal, chylomicrons are formed to carry fat and cholesterol from the intestine into the bloodstream. Chylomicrons are also necessary for the absorption of certain fat-soluble vitamins, such as vitamin E and vitamin D.

Chylomicrons are made up mostly of various types of fats (lipids); they also contain some proteins, mainly a protein called apolipoprotein B-48 that is produced only in the intestine. Because chylomicrons are made up of lipids and proteins, they are known as lipoproteins. Chylomicrons are released from enterocytes into the bloodstream so the body can use the lipids and fat-soluble vitamins they carry. Sufficient levels of fats, cholesterol, and vitamins are necessary for normal growth and development.

Health Conditions Related to Genetic Changes

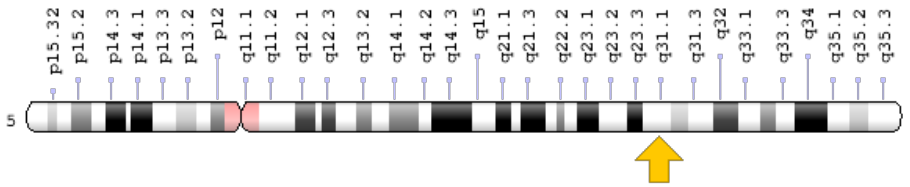
chylomicron retention disease

More than 10 mutations in the *SAR1B* gene have been found to cause chylomicron retention disease. Most of these mutations change one protein building block (amino acid) in the SAR1B protein. Other mutations lead to the production of an abnormally small version of the protein that cannot function properly. Mutations disrupt the SAR1B protein's ability to transport chylomicrons within cells, which prevents chylomicron release into the bloodstream. This retention of chylomicrons prevents dietary fats and fat-soluble vitamins from being used by the body, leading to the nutritional and developmental problems seen in people with chylomicron retention disease.

Chromosomal Location

Cytogenetic Location: 5q31.1, which is the long (q) arm of chromosome 5 at position 31.1

Molecular Location: base pairs 134,601,149 to 134,632,843 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GTP-binding protein Sara
- SAR1 gene homolog B (S. cerevisiae)
- SAR1 homolog B (S. cerevisiae)
- SAR1a gene homolog 2
- SAR1B_HUMAN
- SARA2
- secretion associated, Ras related GTPase 1B

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Alterations of the Exocytic Pathway Lead to Severe Development Defects
  https://www.ncbi.nlm.nih.gov/books/NBK6443/#A75583

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SAR1B%5BTIAB%5D%29+OR+%28SARA2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SAR1, S. CEREVISIAE, HOMOLOG B
  http://omim.org/entry/607690

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SAR1B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SAR1B%5Bgene%5D
- HGNC Gene Family: ARF GTPase family
  http://www.genenames.org/cgi-bin/genefamilies/set/357
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10535
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/51128
- UniProt
  http://www.uniprot.org/uniprot/Q9Y6B6

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